Curating the Clinical Genome (CCG) 2023 Abstract

Title:

Batch ClinVar Submission Support in ClinGen's Variant Curation Interface (VCI)

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Abstract:

The NIH-funded Clinical Genome Resource Consortium Variant Curation Interface (ClinGen VCI) is a global, open-source variant classification platform for supporting the application of evidence criteria and classification of variants based on the ACMG/AMP sequence variant classification guidelines. To facilitate evidence-based improvements in human variant classification, the VCI is publicly available to the worldwide genomics community. The VCI is among a suite of tools developed by ClinGen, and supports an FDA-recognized human variant curation process of ClinGen Variant Curation Expert Panels (VCEPs). ClinGen is expanding to involve more curators and teams of curators working today (affiliations) as part of an increasing scale of activities that will increase genetic variants across a greater number of genes. The variant curation workflow is intended to support dissemination of variant curations into two repositories: the ClinGen Evidence Repository (for approved ClinGen VCEPs) and ClinVar. Support for dissemination of curations into the Evidence Repository from the VCI is an API-driven process via the ClinGen Data Exchange; however, support for ClinVar submission from the VCI has been largely manual to date.

Here we present the first in a series of planned software features to provide a better ClinVar submission experience for VCI users. This batch submission feature allows users to create an active batch of curated variants and then download a preformatted file of the curation data, which can easily be submitted to ClinVar. This provides a faster process for users submitting variant interpretations to ClinVar. It also allows VCI users to annotate which of their curations have been submitted to ClinVar. Future feature plans for ClinVar submission from the VCI include providing VCI users with real-time feedback on the status of their ClinVar submissions, as well as direct API-based submissions from the VCI. These features together will further streamline workflows

for both ClinGen VCEP and non-ClinGen VCI users, and support the ClinGen goal of creating scalable curation workflows to support the clinical genomics community.